

Multi-omics characterization of skeletal muscle in health and disease

Research project for Engineer or Master students

Congenital myopathies are a class of rare genetic diseases characterized by muscle weakness from birth and abnormalities in skeletal muscle biopsies. There is currently no curative treatment for these diseases as the understanding of the pathological mechanisms remains limited.

Our team previously identified the genetic basis of several myopathies and characterized corresponding and faithful mouse models. Defects in skeletal muscle structure and function were observed but their molecular bases are unknown.

Using **multi-omics approaches (single nuclei and bulk transcriptomic, proteomic, metabolomic)**, we have begun to unravel molecular pathways that appear altered in several myopathies. The student will proceed to the analysis of the omics data generated to identify the pathological signatures. He/she will then combine these different multi-omics levels into an **integrated view** of these pathologies using different informatic tools and pipelines. Wet-bench validation will be done by the student or other team members through alternative methods ranging from western blotting, quantitative PCR, ELISA assays and metabolite dosing.

Overall, the student will propose and confirm the main molecular pathways implicated in different myopathies. These pathways will be candidate for genetic or/and pharmacological manipulation to validate novel therapeutic proof-of-concept.

Candidate profile: you are a highly motivated and talented student (Engineer final year or Master 2), with strong interest in understanding the mechanisms of human diseases. Proven expertise in bioinformatics and big data analysis. English is the communication language in the team.

The internship will take place in Strasbourg at the **IGBMC** institute that is one of the main European centers in Biomedical research and offers a unique environment with 50 research teams, 45 different nationalities and a dozen of state-of-the-art platforms.

Strasbourg is a cosmopolitan city in a beautiful countryside, close to Germany and less than 2 hours from the center of Paris by train.

Applications: Alix SIMON (<u>simonal@igbmc.fr</u>) and Jocelyn LAPORTE (<u>jocelyn@igbmc.fr</u>) Ph +33(0)388653415 www.igbmc.fr/laporte